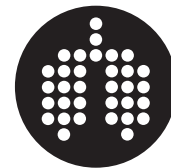


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"Clinical phenotypes of Italian and Spanish patients with α_1 -antitrypsin deficiency"

1. Which of the following statements concerning α_1 -antitrypsin deficiency (AATD) is true?

- It is associated with increased risk of pulmonary emphysema. It is associated with increased risk of bronchial asthma. It is associated with increased risk of pneumonia. It is associated with increased risk of lung cancer

2. Which of the following statements concerning AATD is false?

- There are more than 120 genetic variants of AAT. The most frequent deficient allele is called Z. The main target of the AAT is neutrophil elastase. The most frequent allele characterised by normal serum levels of AAT is called S.

3. Which of the following statements concerning clinical characteristics of patients with AATD is true?

- Patients with emphysema and AATD use to be older at onset of symptoms than patients with smoking COPD. The most frequent reason for AATD diagnosis is liver disease. Around 20% of patients with AATD have history of recurrent pneumonia. More than 50% of patients with AATD have bronchiectasis.

4. Of the following statements concerning clinical characteristics of patients with AATD is false?

- Emphysema is the most frequent pulmonary manifestation of AATD. Around 30–40% of patients with pulmonary disease and AATD in Spain and Italy are receiving augmentation therapy with intravenous AAT. Patients with AATD and overlap with asthma have more preserved FEV1 (% predicted) compared with other phenotypes. Patients with PI*SZ phenotype have more preserved lung function compared with PI*ZZ

5. Which of the following statements concerning AATD registries is false?

- Registries are needed to collect enough information for rare diseases such as AATD. Registries of AATD include heterozygous PI*MZ. Spanish and Italian registries use the same database. Spanish and Italian registries were founded in the 1990s.